Letter to the Editor

Geleophysic Dysplasia and Myhre Syndrome: Reply to Luis E. Figuera

To the Editor:

We thank Figuera [1996] for pointing out Myhre syndrome [Myhre et al., 1981] as a possible differential diagnosis for our patient [Rosser et al., 1995]. Several overlapping manifestations were present in our patient. He had low birth weight, decreased joint mobility, and hearing impairment. While Myhre syndrome should be considered, several findings in our patient favored the diagnosis of geleophysic dysplasia. When first examined, the most striking abnormality he had was thickened, edematous skin. He had the small hands and feet and "tiptoe" gait characteristic of patients with geleophysic dysplasia. He had no evidence of muscle hypertrophy, which one would expect with Myhre syndrome. His facial appearance was that of geleophysic dysplasia, and echocardiography showed him to have a thickened mitral valve. It has been shown [Shohat et al., 1990] that there may be variations in severity of cardiac disease in geleophysic dysplasia. In Myhre syndrome, cardiovascular abnormalities range from structural and conductive abnormalities of the heart to systemic hypertension [Myhre et al., 1981; Soljak et al., 1983; García-Cruz et al., 1993] and thickened mitral valves have not been described. Our patient also had hepatomegaly, which has not been described in Myhre syndrome. While there was advanced paternal age at birth, as seen in Myhre syndrome, the possibility of an autosomal recessive condition cannot be excluded. Overall, we conclude that his clinical picture was more consistent with the diagnosis of geleophysic dysplasia.

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